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Usher's syndrome – case report

Zespół Ushera – opis przypadku

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Summary: The aim of this study is to present a case of coincidence of sensorineural hearing loss with chronic recurrent bilateral cystoid macular oedema in a 32-year-old woman, who was admitted to the clinic for deterioration of visual acuity of four months' duration. The patient gave a history of hearing loss for 29 years.
Visual field examination disclosed peripheral ring scotoma. Electrophysiological examination was performed: pattern visual evoked response was within normal limits and electroretinogram displayed diminished both photopic and scotopic response. As ophthalmoscopy demonstrated no pigment in the fundus of the eye, the findings were consisted with diagnosis of retinitis pigmentosis sine pigmento. The presence of loss of hearing indicated the necessity of performing the genetic examination for Usher's syndrome.
In order to establish a final diagnosis of Usher's syndrome genetic examination must be performed, but family history is relevant. Early investigation for Usher's syndrome in children with sensorineural hearing impairment is of a great significance. The patient may develop symptoms of retinitis pigmentosa in second or even third decade of his life. The necessity of thorough investigation for detecting other systemic abnormalities should be emphasized.
There is no effective treatment of this syndrome. A child with Usher's syndrome requires a comprehensive care of different medical specialties. Psychological, educational and sociological attitude is also of a great importance in the child development.

Key words: Usher's syndrome, bilateral cystoid macular oedema, sensorineural hearing loss, genetic disorder.
Słowa kluczowe: Zespół Ushera, obuoczny torbielowaty obrzęk plamek, niedosłuch typu odbiorczego, choroby genetyczne.

Purpose

The aim of this study is to present a case of coincidence of sensorineural hearing loss with chronic recurrent bilateral cystoid macular oedema in a 32-year-old woman.

Clinical description

In February 2006, a 32-year-old woman was admitted to the clinic for deterioration of visual acuity of four months' duration. Bilateral central retinitis was diagnosed in outpatient department. The patient gave a history of hearing loss for 29 years. It was probably caused by administration of gentamycin in neonatal period. Audiological examination disclosed sensorineural hearing loss. Physical examination showed decreased visual acuity to 0.4 in right eye and up to 0.6 in left eye. Anterior segment was normal. Indirect ophthalmoscopy showed pink, of a distinct boarders optic disc, c/d ratio 0.4, constricted vessels, attached, pink retina, cystoid macular oedema and lattice degenerations in peripheral part of retina. Fluorescein and indocyanine green angiography was non-contributory. OCT examination proved the presence of cystoids macular eodema. The central retinal thickness is 350 μm in right eye and 362 μm in left eye. Systemic, connective tissue and animal borne diseases diagnostic investigations were non- contributory. The patient was administered Encorton in daily doses of 35 mg, Diuramid in two equal doses of 250 mg and Kalipoz 3 tablets daily. The treatment was initiated orally. The patient's uneventful recovery and regression of macular eodema were fallowed by gradual reduction of the medicaments dosage. In a result recurrence of the symptoms were observed after a month. After readministration of the Diuramid and Kalipoz the symptoms and signs subsided rapidly. Attempt of discontinuing of the treatment resulted in recurrence of the macular oedema. For next 18 months visual acuity was between 0.3 and 1.0, the central macular thickness was between 201 μm and 446 μm depending on oral trealtment. In December 2007 Lucentis in a dose of 0,5 mg was administered intravitreously in the right eye which caused complete reduction of macular oedema with increase in visual acuity up to 1.0 of two and half months' duration. In January 2008 Avastin in a dose of 1.25 mg was injected intravitreously in the left eye. Reinjection of Lucentis in the right eye was performed in March 2008. Both the injections caused regression of oedema of macula. As the rise in intraocular pressure up to 60 mm Hg was noted during a treatment of both medications, the glaucoma investigations were ordered. Phasing did not revealed anything specific: right eye: 12-16 mm Hg, left eye: 12-18 mmHg. Visual field examination disclosed peripheral ring scotoma. Electrophysiological examination was decided to perform: pattern visual evoked response was within normal limits and electroretinogram displayed diminished both photopic and scotopic response. The results of this examination are probably suggestive of retinitis pigmentosa. As ophthalmoscopy demonstrated no pigment in the fundus of the eye, the findings were consisted with diagnosis of retinitis pigmentosis sine pigmento. The presence of loss

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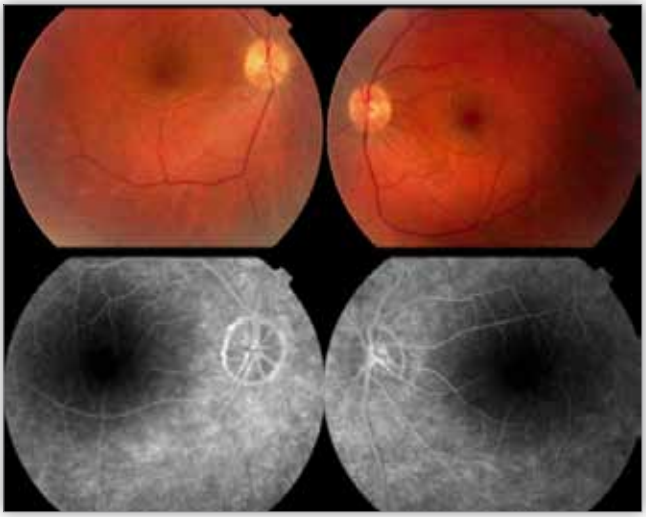

Fig. 1. Colourful eye fundus photograph and fluorescein angiography.
Ryc. 1. Zdjęcie kolorowe dna oka i angiografia fluoresceinowa.

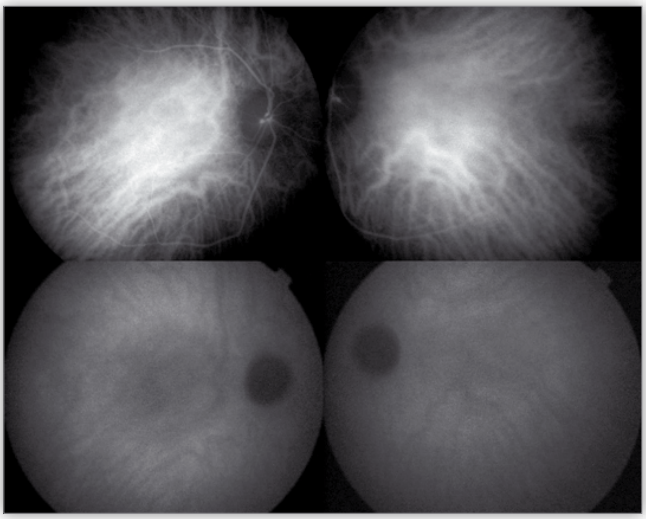

Fig. 2. Indocyanine green angiography.
Ryc. 2. Angiografia indocyjaninowa.

of hearing indicated the necessity of performing the genetic examination for Usher's syndrome (Fig. 1-4).

Usher's syndrome is a disorder inherited in an recessive pattern. It is characterised by the presence of sensorineural hearing loss and retinitis pigmentosa. Apart from that other disorders may be developed (1).

The three types of the Usher's syndrome are distinguished:

- Type 1 – profound inborn bilateral deafness, difficulties in maintaining the balance due to disturbance of vestibular system function, retardation in developing motor skills, speech impediment, retinitis pigmentosa;
- Type 2 – moderate to severe hearing loss with no balance disturbances, retinitis pigmentosa;
- Type 3 – normal hearing at birth with progressive hearing loss, vestibular dysfunction may occur (2,3).

The presence of retinitis pigmentosa in Usher's syndrome cause the following symptoms: deterioration of scotopic vision, restricted visual field by peripheral scotomas (tunnel vision), diminished contrast sensitivity, decreased visual acuity due to the presence of posterior subcapsular cataract in advanced stages

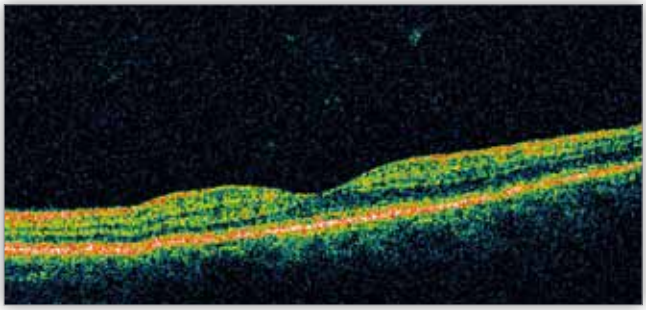

Fig. 3a. OCT after treatment.
Ryc. 3a. OCT po leczeniu.

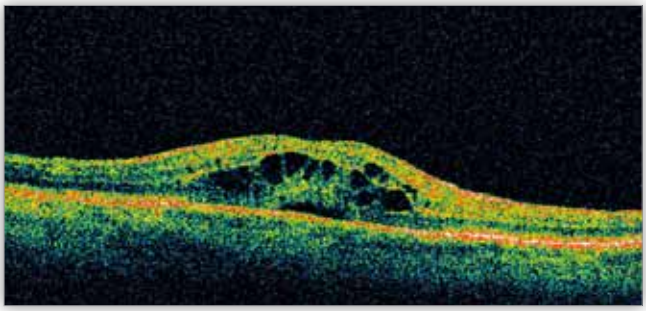

Fig. 3b. OCT before treatment.
Ryc. 3b. OCT przed leczeniem.

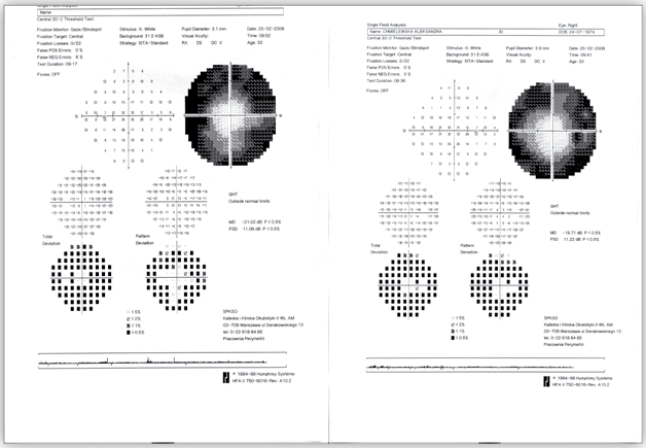

Fig. 4. Visual field.
Ryc. 4. Pole widzenia.

of the disease. Direct ophthalmoscopy showes vessel constriction, pallor of the optic disc, clumps of pigment in peripheral retina. Histophatological examination discloses excessive lipid accumulation in retina as well as in Corti's organ (4,5).

Epidemiology

Carrier state rate of genes causing Usher's syndrome amounts 1: 70 in human population. As results of autosomal recessive disorders the chance for inheritance a disease is 25%.

The Usher's syndrome results from chromosomal mutations of the following genes:

- Type 1: CDH 23, CLRN1, GPR 98, MYO7A, PCDH15, USH1C, USH1G and USH2A;
- Type 2: USH2A and GPR78 (also called VLGR1) and DFNB31;
- Type 3: CLRN1 (1,6,7,8).

Usher's syndrome is responsible for 3-6% children's deafness. It occurs in 4 to 100 000 cases. II type of Usher's syndrome is most frequent. The third type appears in only small number of cases. The incidence of this syndrome like other recessive diseases correlates with a high degree of relation (9).

Diagnosis

In order to establish a final diagnosis of Usher's syndrome genetic examination must be performed. Hearing loss accompanied by retinitis pigmentosa is not sufficient for Usher's syndrome diagnosis, to be established. The following examinations should be performed: ophthalmoscopy, visual field test to detect peripheral vision defect, an electroretinogram (ERG) (10,11,12).

In case of suspicion of the genetically related hearing loss with the presence of retinitis pigmentosa an accurate physical systemic examination should be performed especially taking into account the evidence of dysmorphy. Family history is relevant. Early investigation for Usher's syndrome in children with sensorineural hearing impairment is of a great significance. The patient may develop symptoms of retinitis pigmentosa in second or even third decade of his life. The necessity of thorough investigation for detecting other systemic abnormalities should be emphasized (13,14,15).

Conclusions

There is no effective causal treatment of this syndrome.

A child with Usher's syndrome requires a comprehensive care of different medical specialties. Psychological, educational and sociological attitude is also of a great importance in the child development (14,16,17,18).

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